PCCA gene

propionyl-CoA carboxylase alpha subunit

Normal Function

The *PCCA* gene provides instructions for making part of an enzyme called propionyl-CoA carboxylase, specifically, the alpha subunit of this enzyme. Six alpha subunits come together with six beta subunits (produced from the *PCCB* gene) to form a functioning enzyme. The alpha subunit also includes a region for binding to the B vitamin biotin.

Propionyl-CoA carboxylase plays a role in the normal processing of proteins. It is responsible for a particular step in the breakdown of several protein building blocks (amino acids) called isoleucine, methionine, threonine, and valine. Propionyl-CoA carboxylase also helps break down certain types of lipids (fats) and cholesterol. First, several chemical reactions convert the amino acids, lipids, or cholesterol to a molecule called propionyl-CoA. Using biotin, propionyl-CoA carboxylase then converts propionyl-CoA to a molecule called methylmalonyl-CoA. Additional enzymes break down methylmalonyl-CoA into other molecules that are used for energy.

Health Conditions Related to Genetic Changes

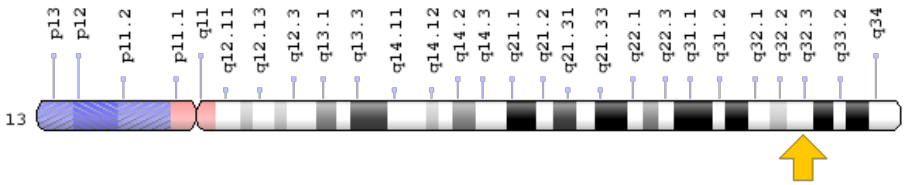
propionic acidemia

More than 45 mutations in the *PCCA* gene have been identified in people with propionic acidemia. These mutations include changes in single DNA building blocks (nucleotides) and insertions or deletions of genetic material in the *PCCA* gene. *PCCA* mutations prevent the production of functional propionyl-CoA carboxylase or reduce the enzyme's activity. The altered or missing enzyme is unable to process certain parts of proteins and lipids properly. As a result, propionyl-CoA and other potentially harmful compounds can build up to toxic levels in the body. This buildup damages the brain and nervous system, causing the serious health problems associated with propionic acidemia.

Chromosomal Location

Cytogenetic Location: 13q32.3, which is the long (q) arm of chromosome 13 at position 32.3

Molecular Location: base pairs 100,089,015 to 100,530,437 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- PCCA_HUMAN
- PCCase alpha subunit
- propionyl CoA carboxylase, alpha polypeptide
- propionyl-CoA:carbon dioxide ligase alpha subunit
- propionyl Coenzyme A carboxylase, alpha polypeptide

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Propionyl-CoA carboxylase deficiency blocks the biotin- and ATP-dependent conversion of propionyl-CoA to methylmalonyl-CoA
  https://www.ncbi.nlm.nih.gov/books/NBK27933/#A3116

GeneReviews

- Propionic Acidemia
  https://www.ncbi.nlm.nih.gov/books/NBK92946

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PCCA%5BTIAB%5D%29+OR+%28propionyl+Coenzyme+A+carboxylase%5BTIAB%5D%29%29+OR+%28propionyl-CoA%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- PROPIONYL-CoA CARBOXYLASE, ALPHA SUBUNIT
  http://omim.org/entry/232000

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PCCA.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PCCA%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8653
- Kraus Lab at the University of Colorado Health Sciences Center
  http://www.ucdenver.edu/academics/colleges/medicalschool/programs/kraus/Pages/home.aspx
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5095
- UniProt
  http://www.uniprot.org/uniprot/P05165

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